Table I Imbalances over 1 Mb detected by a- CGH/ QF-PCR

Imbalance	Number	
QF		
Turner	53	Whole chromosome
Polyploidy	50	Triploid/Tetraploid
Trisomy 18	37	Whole chromosome
Trisomy 21	34	Whole chromosome
Trisomy 13	23	Whole chromosome
Mole	4	
Klinefelter	2	Whole chromosome
Array CGH		
Duplication	19	
Trisomy 22	15	Whole chromosome
Deletion and duplication	14	Unbalanced translocation
Trisomy 15	13	Whole chromosome
Interstitial Deletion	12	Interstitial Deletion
Trisomy 16	9	Whole chromosome
Trisomy 7	5	Whole chromosome
Terminal Deletion	4	Terminal Deletion
Trisomy 9	4	Whole chromosome
Trisomy 4	2	Whole chromosome
Trisomy 8	2	Whole chromosome
supernumerary marker chromosome	2	supernumerary marker chromosome
Mosaic loss	2	Whole Chromosome
Trisomy 11	1	Whole chromosome
Trisomy 19	1	Whole chromosome
Trisomy 20	1	Whole chromosome
Smith Magenis	1	
Di George	1	
Mosaic Deletion	1	Partial
Two Deletions	1	

Table II Samples with imbalance under 1 Mb

Case	Array CGH results	Size	Sex	Couple Related	Other findings	Interpretation
827	*arr[GRCH37]19q13.31(48046129-48221671)x1.mat*	175 Kb	Female	No	Maternal	Likely Benign
999	*arr[GRCH37] Xp21.1(931593574-31896576)x1.mat*	303 Kb	Male	Yes	Maternal, compatible with Duchenne	Pathogenic
1075	*arr[GRCH37]2q13(110862507-110964708)x1*	102 Kb	Female	Yes		VOUS*
1183	*arr[GRCH37] 16p13.11(14910228-16311041)x3.mat* *arr[GRCH37]17q12(31998155-32922936)x3.pat*	1.4 Mb and 924 Kb	Male	No	Macrocephaly, Metaphyseal dysplasia, shortening of long bonesFGFR3 Mutation: **c.1138G.A(p.Gly380Arg) was confirmed**	Benign
1254	*arr[GRCH37]16p13.3(6266327-6735636)X1*	469 Kb	Female	No		VOUS
1325	*arr[GRCH37] 22q11.21(20708912-21440485)x3*	731 Kb	Female	No		VOUS
1351	*arr[GRCH37]Xq26.2(132265298-132675323)x1*	410 Kb	Male	No	Amnion: 46,XY	VOUS
1566	*arr[GRCH37]3p26.1(4261189-4420643)x60*	160 Kb	Male	Yes	Parents are both carrier, Next offspring was affected, compatible with Multiple Sulfatase Deficiency	Likely pathogenic
1869	*arr[GRCH37] 5p13.2p13.2(36520895-37187880)x3*	677 Kb	Male	Yes	Partial absence of corpus callosum, coarse face, flexion contracture of elbow, knee and left wrist, long fingers, Compatible with 5p13 microduplication	Pathogenic
2064	*arr[GRCH37]16p13.11(15534128-16276115)x1*	742 Kb	Male	Yes		VOUS

***VOUS: Variant of uncertain significance**

Figure I Material and methods chart

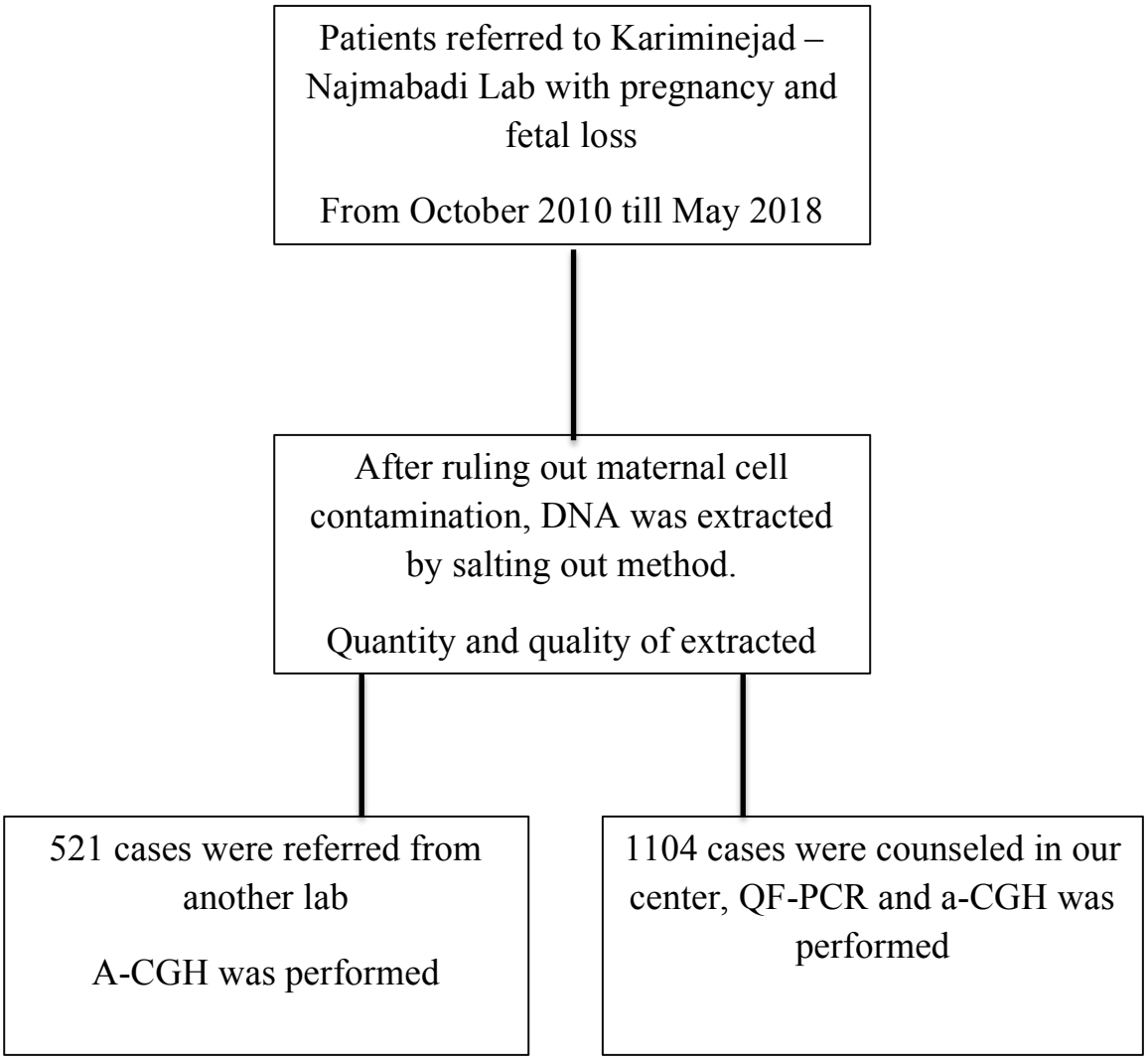

Figure II Detection rate in each semester in related and unrelated cases